Al collegio docenti del Dottorato in Medicina Molecolare

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Ciclo XXXVII Tutor: Prof.ssa Betti Giusti

Dottorato di ricerca a tema vincolato: Medicina molecolare in ambito cardiovascolare e aterotrombotico: utilizzo di tecnologie "omiche" e approcci di biomedicina computazionale (gestione di BigData) per la comprensione delle basi genetico-molecolari e lo sviluppo di strategie diagnostiche personalizzate

Attività scientifica svolta nel 1° anno di Dottorato, Anno Accademico 2021/2022

I attended my first year of PhD at the Advanced Molecular-Genetic Laboratory of Center for Atherothrombotic Diseases, Dpt. Experimental and Clinical Medicine, University of Florence. My activity mainly focused on atherotrombotic and cardiovascular diseases study. Part of the research activities were dedicated on the study of relationship between viral infections and cardiovascular complications; in particular, the research has focused on identification of genetic variants and/or genetic profiles associated with severity of COVID19 disease and thrombotic events susceptibility. A targeted Next Generation Sequencing (NGS) was performed by libraries preparation according to HaloPlex HS Target Enrichment System protocol(Agilent Technologies) and Illumina MiSeq, on a cohort of n=40 patients hospitalized at AOUCareggi; genetic analysis included a sequencing panel of 11 genes known to be involved in coagulation processes. Twenty-nine rare (MAF≤1%) variants have been identified at the heterozygous state in 24/40 patients. A higher prevalence of rare missense variants with potential pathogenic prediction in patients transferred in intensive care unit or dead (26.7%) was observed than in ordinary ward patients (8%), suggesting how individual genetic profiles may participate in modulating susceptibility to SARS-CoV-2 infection. RT-PCR genotyping analysis of factor V Leiden (rs6025) and G20210A F2 (rs1799963) polymorphisms, involved in common thrombophilia, has been also conducted in n=994 patients with COVID19; the presence of common genetic factors does not seem to indicate a significant risk modulation of thromboembolic complications.Moreover, 6 patients with Vaccine-induced thrombocytopenia, a prothrombotic syndrome observed in a small number of individuals who received the adenoviral vector-based vaccine Vaxzevria (astrazeneca) was performed by WES approach showing a wide number of variants in molecular pathways involved in integrin signaling, thrombocytopenia and platelet aggregation/activation processes.
At the same time, the study of the genetic bases of hereditary thoracic aortopathies was conducted. We published a review which summarizes the state of the art of the literature and our research on thoracic aortic aneurysm/dissection, with particular reference to current and future role of molecular-genetic testing.

• Pubblicazioni scientifiche:

-De Cario R, Giannini M, Cassioli G, Kura A, Gori AM, Marcucci R, Nistri S, Pepe G, Giusti B, Sticchi E. Tracking an Elusive Killer: State of the Art of Molecular-Genetic Knowledge and Laboratory Role in Diagnosis and Risk Stratification of Thoracic Aortic Aneurysm and Dissection. Diagnostics (Basel). 2022 Jul 22;12(8):1785. doi: 10.3390/diagnostics12081785. PMID: 35892496; PMCID: PMC9329974.

•Partecipazione a webinar, seminari e congressi:
-DENOThe "Role of hyponatremia on tumoral proliferation and invasiveness and possible new anti-cancer strategies"; Giada Marroncini, 30/11/2021
-DENOThe "Use of modern imaging techniques in teaching anatomy and research"; Petr Zach, 19/01/2022
-EAS Lipid Clinic Webinar “Le linee guida per le dislipidemie: presente e futuro”; 5/11/2021
-NeuroWebinar: “Functional dynamics of chromatin topology in human cardiogenesis and disease” 18/02/2022
-IPAM 2022, 3Days for 3Rs: Replacement, Reduction, Refinement; 07-14-21/02/2022
-DENOThe “Exploring the genomic and epigenomic landscape of Acute Myeloid Leukemia with Nanopore sequencing”,Alberto Magi,20/04/2022
-Training Illumina, Next Seq 2000 Costumer Training, Licia Iaccarino, 26-27/04/2022
-Training TermoFisher Scientific 2500Dx, Sequencing and Instrument operation course, Fabio Raffaldi, 13-14/07/2022
-Lipidologia, medicina interna e cardiologia: quale sinergia nella gestione dell’ipercolesterolemia severa? Pisa, 24/11/2021
-Cardiology Grand Rounds 2021 Firenze, 02/12/2021

• Abstracts:
- Marco Giannini, Elena Sticchi, Tommaso Capezzuoli, Rosina De Cario, Samuele Suraci, Giulia Cassioli, Sara Neroni, Angela Antonietta Rogolino, Martina Berteotti, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Identification of rare coagulation variants and screening of thrombophilic polymorphisms (f2 rs1799963, f5 rs6025) in COVID-19 patients. CORSO NAZIONALE SISET 2022, 02/11-05/11 2022.
-Giulia Barbieri, Tommaso Capezzuoli, Marco Giannini, Giulia Cassioli, Francesca Cesari, Rossella Marcucci, Anna Maria Gori, Betti Giusti, Elena Sticchi. Lpa kringle iv type 2 detection: qpcr or ddpcr? CORSO NAZIONALE SISET 2022, 02/11-05/11 2022
-Rosina De Cario, Elena Sticchi, Ada Kura, Tommaso Capezzuoli, Marco Giannini, Giulia Cassioli, Giulia Barbieri, Samuele Suraci, Anna Maria Gori, Martina Berteotti, Angela Antonietta Rogolino, Rossella Marcucci, Betti Giusti. Individual susceptibility to sars-cov-2 infection: role of the host’s virus entry machinery genetic profiles. CORSO NAZIONALE SISET 2022, 02/11-05/11 2022.
-Angela Rogolino, Francesca Cesari, Anna Maria Gori, Betti Giusti, Bandinelli Brunella, Elena Sticchi, Alessia Bertelli, Marco Giannini, Daniela Poli, Elena Lotti, Lucia Mannini, Rossella Marcucci. Vaccine-induced thrombotic thrombocytopenia (VITT): evaluation of immunologic and functional tests on an Italian case series. CORSO NAZIONALE SISET 2022, 02/11-05/11 2022.
-Tommaso Capezzuoli, Rosina De Cario, Elena Sticchi, Marco Giannini, Rebecca Orsi, Lapo Squillantini, Samuele Suraci, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Vaccine induced thrombotic thrombocytopenia: evaluation of genetic susceptibility throught whole exome sequencing. CORSO NAZIONALE SISET 2022, 02/11-05/11 2022.